Recombinant Human ATXN7, His-tagged

Cat. No. ATXN7-3690H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Ataxin-7 (ATXN7)
Species	Human
Source	E.Coli/Yeast
ProteinLength	892
Description	The autosomal dominant cerebellar ataxias (ADCA) are a heterogeneous group of neurodegenerative disorders characterized by progressive degeneration of the cerebellum, brain stem and spinal cord. Clinically, ADCA has been divided into three groups: ADCA types I-III. ADCAI is genetically heterogeneous, with five genetic loci, designated spinocerebellar ataxia (SCA) 1, 2, 3, 4 and 6, being assigned to five different chromosomes. ADCAII, which always presents with retinal degeneration (SCA7), and ADCAIII often referred to as the "pure" cerebellar syndrome (SCA5), are most likely homogeneous disorders. Several SCA genes have been cloned and shown to contain CAG repeats in their coding regions. ADCA is caused by the expansion of the CAG repeats, producing an elongated polyglutamine tract in the corresponding protein. The expanded repeats are variable in size and unstable, usually increasing in size when transmitted to successive generations. This locus has been mapped to chromosome 3, and it has been determined that the diseased allele associated with spinocerebellar ataxia-7 contains 38-130 CAG repeats (near the N-terminus), compared to 7-17 in the normal allele. The encoded protein is a component of the SPT3/TAF9/GCN5 acetyltransferase (STAGA) and TBP-free TAF-

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	containing (TFTC) chromatin remodeling complexes, and it thus plays a role in transcriptional regulation. Alternative splicing results in multiple transcript variants.
Form	This item requires custom production and lead time is between 5-9 weeks. We can custom produce according to your specifications.
Purity	>90%
Notes	Small volumes of ATXN7 recombinant protein may occasionally become entrapped in the seal of the product vial during shipment and storage. If necessary, briefly centrifuge the vial on a tabletop centrifuge to dislodge any liquid in the container`s cap. Certain products may require to ship with dry ice.
Storage	Store at -20 degree C. For extended storage, store at -20 or -80 degree C.
Storage Buffer	PBS pH 7.4, 50% glycerol
Warning	This product is for research use only. Not for use in diagnostic or therapeutic procedures.

GENE INFORMATION

Gene Name	ATXN7 ataxin 7 [Homo sapiens]
Official Symbol	ATXN7
Synonyms	ATXN7; ataxin 7; SCA7, spinocerebellar ataxia 7 (olivopontocerebellar atrophy with retinal degeneration); ataxin-7; ADCAII; OPCA3; spinocerebellar ataxia type 7 protein; SCA7; FLJ17787;
Gene ID	6314

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mRNA Refseq	NM_000333
Protein Refseq	NP_000324
MIM	607640
UniProt ID	O15265
Chromosome Location	3p21.1-p12
Function	chromatin binding; protein binding; zinc ion binding;

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